Project title	**Fabry Family Screening** A collaborative working project between Amicus Therapeutics UK Operations and University Hospitals Birmingham NHS Foundation Trust (UHB)
Project rationale	**Project Rationale:** • Fabry disease is an X-linked lysosomal storage disorder which manifests as multi-organ accumulation of glycosphingolipids due to enzyme deficiency of alpha-Galactosidase A. • A pedigree analysis should be carried out in each patient with confirmed Fabry disease. • Due to multiple reasons including time and experience this care is not always consistent and can lead to long waiting times for patients to be seen resulting in delays in diagnosis and treatment and ultimately patient outcomes. • It is therefore important to improve Fabry family screening and the potential of specialist intervention.
Project objectives	**Project objectives** • Support priority one of the UK Rare Disease Framework - Helping patients get a final diagnosis faster • 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 • Demonstrate an improvement in the coordination of care provided for Fabry patients and their families - Leading to better patient experience • Demonstrate the value of genetic counsellors to facilitate the improvement in screening uptake. **Potential patient Benefits** • Availability and information about Fabry disease inheritance patterns. • Genetic screening for family members at risk of Fabry Disease. • Helping patients get a final diagnosis faster • Earlier access to clinically appropriate treatment for newly diagnosed patients. **Potential NHS Benefits** • Improved co-ordination of care for Fabry patients, resulting in earlier diagnosis and thereby compliance with Trust and UK Rare Diseases strategy. • Develop a professional relationship with industry partners in a transparent and collaborative manner • Leverage professional, Trust and industry expertise, with active involvement of patient societies, in such a way as to enhance availability of mapping and screening in a sustainable, fully funded way. • Increase in appropriately treated patients. • Improved patient satisfaction **Potential Amicus Benefits** • Better understanding of the barriers to uptake in Fabry genetic counselling and screening. • Participation in the implementation and ongoing development of the UK Rare Disease Framework. • While not a deliverable pursuant to the project, as a consequence of patients' diagnosis, a proportion of these patients may be clinically eligible for treatment with Amicus's product for Fabry disease.
Project period	Q4 2022 – Q1 2026 The project has been extended and will now run to Q1 2027 **Reason for extension** To focus on a cohort of patients who have not yet engaged in the project to truly understand the reasons for this and if proactive contact aligned with an agreed consensus approach has a positive impact on engagement as there is currently limited data in this area.
Financial arrangements	The project will be supported by a pooling of skills and resources from both Amicus and UHB, with a direct financial contribution provided by Amicus of approx. £163,000
Plan for publication	Clinical Medicine Journal, WORLD*Symposium* 2026, WORLD*Symposium* 2027, The BIMDG Symposium 2026, AGNC (Association of Genetic Nurses and Counsellors) Conference 2027
Contact details	Laura Clark General Manager Amicus Therapeutics lrclark@amicusrx.com Professor Rick Steeds Consultant Cardiologist UHB
Summary of the roles and responsibilities of each party	Roles and responsibilities are agreed and reviewed at monthly steering group meetings