

Project title	Fabry family screening A collaborative working project between Amicus Therapeutics UK Operations and Salford Care Organisation (Part of Northern Care Alliance)
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 factors including time and experience this care is not always consistent and can lead to long waiting times for patients 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 The overall objectives of this project are to: - 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 a genetic practitioner to facilitate the improvement in screening uptake. Potential patient Benefits - Availability and information about Fabry disease inheritance patterns. 1:1 appointment with a genetic practitioner. - 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. - IMD service improvement, through wider engagement and empowerment of patients/families/carers of Fabry patients at all stages of the disease. - Develop a professional relationship with industry partners in a transparent and collaborative manner - Identify and address any health inequalities which may exist in the service - 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	Q2 2025 – Q2 2027
Financial arrangements	The project will be supported by a pooling of skills and resources from both Amicus and Salford Care Organisation, with a direct financial contribution provided by Amicus of £107,000
Plan for publication	WORLDSSymposium 2027, The BIMDG Symposium 2027, AGNC (Association of Genetic Nurses and Counsellors) 2027
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Summary of the roles and responsibilities of each party	Roles and responsibilities are agreed and reviewed at monthly steering group meetings