Project Outcomes Report

Project name: **Flash Mob Audit: Improving knowledge of Fabry Disease**

Project partners: Royal Free London NHS Foundation Trust and Amicus Therapeutics UK Ltd

Duration: February 2022 – February 2023

Project overview:

The project aim was to support cardiology education in Fabry disease and to implement learnings through live audits – which support patients and clinicians in addressing the diagnostic odyssey in Fabry disease.

Patient Benefits

- Reduced length of time to diagnosis of Fabry disease through improved knowledge of health care professionals.
- Better experience for patients and improved confidence of HCPs.
- Improved management of Fabry disease through improved knowledge of HCPs.

NHS Benefits

- Upskilled cardiology workforce in an area where education is limited supporting implementation of the 2021 UK Rare Disease Framework.
- Quality improvement through education and implementation of the audit learnings to reduce time to diagnosis of Fabry disease for patients with cardiac symptoms.
- Improved collaboration and communication across NHS services for Fabry patients.
- Improved experience of patients with Fabry disease through improved HCP education.
- Supporting new ways of implementing clinical audit against current guidelines in the NHS.
- Increased collaboration between trainees through generation of educational networks.

Amicus Benefits

- Better holistic care and in turn outcomes for Fabry patients supporting our mission to improve the lives of patients with rare disease.
- Potential expansion of the Fabry patient population as a result of this project improving identification of Fabry disease and reducing time to diagnosis.

Key Deliverables

- Develop and deliver an educational program to the national network of cardiology doctors in training.
- Run national 'Flash Mob Audit' day – which involved anonymous data collection from trust cardiology datasets.
- Support participants in the analysis and review of the data – and feed back to participants with key outcomes to implement quality improvement locally.
- Write up data outcomes for publication and presentation in various relevant fora.

Collaborative Working project: Royal Free London NHS Foundation Trust and Amicus Therapeutics UK
February 2022

Project outcomes:

- Cardiology and Fabry focussed education was delivered to 17 participants across 11 targeted cardiology sites.
- >140 cardiology patients were reviewed through live audits of trust cardiology datasets.
- The mean data capture time was 4 minutes per clinic.
- The datasets were audited against a combination of the British Inherited Metabolic Disease Group – Fabry guidelines and the European expert consensus statement on therapeutic goals in Fabry disease by Wanner et al.
- The project identified 44% missed screening opportunities for Fabry disease.
- Key outcomes and improvement opportunities fed back to participants for local implementation.
- Review of Flash Mob Audit outcomes and write up / publications: Abstract presented at 2023 British Society of Cardiology conference and publication in the British Medical Journal in June 2023.

Patient Benefits

- The project supported the earlier identification of Fabry disease patients.
- The project aligned with the UK Rare Disease Framework priority to reduce the time to diagnosis in rare disease.

NHS Benefits

- Anonymised combined data was shared back to the Royal Free London NHS Foundation Trust. The data was collated, analysed and then discussed at a national learning event.
- The outcomes were published to increase awareness of Fabry disease and likely indicators of early diagnosis.

Amicus Benefits

- Both parties have benefitted from the project and there has been a strong and trusting relationship throughout the collaboration.
- The pilot in Cardiology has met the criteria to scale into other conditions related to Fabry disease symptoms.

Conclusions and learnings:

- The project has involved considerable and equal contributions from both parties – in line with the original proposal.
- The project had clear and measurable outputs – as the audits were completed with results and actions fed back to the participating centres.
- The abstract presentation and the journal submission were both accepted.
- The project underscores the pressing need to increase awareness for Fabry Disease in patients presenting with Left Ventricular Hypertrophy (LVH).
- Comprehensive cardiac imaging should be a standard protocol for those with Electrocardiography (ECG) abnormalities, ensuring no cases of LVH, potentially indicative of Fabry Disease, go unnoticed. Especially as some of the hallmarks of Fabry disease such as late

gadolinium enhancement of the basal inferolateral wall are only delineated through cardiac Magnetic Resonance Imaging (MRI).

- We advocate for more extensive educational initiatives targeting healthcare professionals to ensure Fabry Disease remains a top differential in patients with unexplained LVH, ensuring timely intervention and improved patient outcomes.
- Future projects could focus on broadening the sample size and understanding potential barriers to care and access to tests such as blood (alpha-galactosidase A enzyme), and genetic testing, especially among minority groups.
- Our study also highlighted the potential racial and ethnic disparities in the diagnosis. While the majority of patients were of white ethnicity, further research is required to ascertain if there are any barriers to care or diagnostic biases that might affect minority groups.

References:

- UK Rare Disease Framework
  - https://www.gov.uk/government/publications/uk-rare-diseases-framework
- British Medical Journal
  - https://heart.bmj.com/content/109/Suppl_3/A29.2

***To be approved by both parties & hosted on Amicus / NHS websites**