Project Name: Hereditary transthyretin amyloidosis (hATTR) service improvement at the Midlands Amyloidosis Service (MAS)

Partner Organisation: University Hospitals Birmingham (UHB)

Completion Date: September 2024

Outcome Summary:

All objectives of this joint working project were met, including the overarching aim to improve patient pathways and timely access to specialist care for patients with hereditary transthyretin amyloidosis and potentially affected family members. New clinical nurse specialist (CNS) led family screening clinics and pathways were implemented in addition to CNS-led coordination of patient reviews for the MAS and multidisciplinary team (MDT) meetings with National Amyloidosis Centre (NAC), benefitting patients with several types of amyloidosis.

Further, the amyloidosis CNS is now a full-time funded position at UHB.

Key Project Outcomes Data:

The joint working project plan outlined several milestones and activities, whose progress was tracked in regular steering committee meetings. The outcome of primary activities is detailed below:

Main activity 1: Onboarding and training of the CNS in inherited conditions, amyloidosis, and genetic counselling, enhancing the management of patient pathways at UHB.
Outcome measure: The CNS received comprehensive training in amyloidosis, inherited cardiac conditions, and genetic counselling, along with practical experience in generating family pedigree trees and managing heart failure. Overall, this comprehensive onboarding and training approach aimed to empower the CNS to confidently navigate the complexities of patient pathways, ensuring high-quality care and support for patients and families affected by amyloidosis.

Main activity 2: Develop a pathway to identify all MAS index patients with hATTR amyloidosis and invite index patients and first-degree relatives for family cascade screening.
Outcome measure: The CNS developed a letter in collaboration with an hATTR patient advocate to explain and offer cascade screening and genetic counselling to index patients and family members. In independent CNS-led telephone clinics, all identified index patients were reached out to.

Main Activity 3: Perform CNS-led hATTR amyloidosis genetic counselling clinics to initiate clinical or genetic screening (if clinically indicated).
Outcome measure: CNS generated 4-generation family pedigree trees and counselled patients and/or family members for hATTR amyloidosis and associated genetics in CNS-led clinics. Individuals of multiple affected families were referred to genetics for potential pre-symptomatic genetic screening. DPD scintigraphy scans were performed as part of clinical screening.

Main Activity 4: Preparation of patient reviews for consultant-led MAS clinics and MDTs (for patients with AL, hATTR and wtATTR amyloidosis).
Outcome measure: The CNS assisted in preparing patient reviews for MAS clinics and took on the responsibility of coordinating patient reviews for MDTs, ensuring all patient assessment forms were complete for thorough evaluation.

Conclusion:

The joint working project demonstrated the value of CNS-led family counselling clinics in improving timely coordination and referrals for genetic and clinical screening of at-risk family members. CNS oversight of patient reviews for MAS clinics and MDTs further enables efficient evaluation of amyloidosis patients in a changing landscape of amyloidosis care in the UK.

As a result of this successful Joint Working project, the CNS role has become a permanent, full-time position at UHB with the outcomes of the project being shared with her peers.