Executive Summary

The Royal Free London NHS Foundation Trust, National Amyloidosis Centre (NAC) and Alnylam UK (Alnylam) Joint Working Project: hATTR amyloidosis Genetic Counsellor

Executive summary

Alnylam and the NAC will enter a collaboration (Joint Working Agreement (JWA)) to increase the availability of genetic counselling in line with consensus on best practice to (i) all patients diagnosed with a hereditary amyloidosis gene variant ("**Index Patients**") and (ii) relatives of the individuals above, regardless of whether they also have been diagnosed with a gene variant. The genetic counsellor funded under this joint working will provide support and advice to patients and their relatives. This support and advice will primarily relate to family cascade screening, implications for relatives, and information on hereditary amyloidosis disease management and relevant NHS services. The genetic counsellor will not recommend, advise or discuss individual available treatments.

In addition, this JWA will increase the capacity of the NAC to process genetic tests for hereditary amyloidosis gene variants. It is expected that the number of requests for genetic tests for an amyloidogenic gene variant will increase as a result of the increased availability of genetic counselling established by this joint working project. Alnylam will fund an additional lab technician at the NAC to assist with the processing and analysis of the increased number of requests for genetic tests for an amyloidogenic gene variant.

The project will commence in November 2023. The project term is 37 months. A short summary of outcomes and lessons learned will be published collaboratively by the NAC and Alnylam.

Expected benefits to patients, the NAC and Alnylam

Patients

It is expected that patients diagnosed with an amyloidogenic gene variant will benefit from improved experiences through the provision of support, advice and information in relation to the diagnosis and its implications. The families of patients diagnosed with an amyloidosis gene variant will also benefit from support, advice and information regarding the potential implications of this diagnosis and this may lead to earlier diagnosis of family members with hereditary amyloidosis. It is expected that the experience of all individuals who undergo genetic testing for an amyloidogenic gene variant will be improved by ensuring that there are fewer delays in receiving the results of tests.

NAC

The NAC will benefit from an increased ability to meet expert opinion consensus on best practice in relation to hereditary amyloidosis. The NAC will also benefit from an increased capacity to process requests for genetic tests for an amyloidogenic gene variant. In addition to this, the project is expected to provide the NAC with some data to support a business case to be presented to RFL Trust and NHS England for the future funding of a permanent genetic counsellor and for future funding of genetic testing capacity at the NAC.

Alnylam

Alnylam is expected to benefit through the potential earlier diagnosis of hATTR patients.